



B3GLCT gene

beta 3-glucosyltransferase

Normal Function

The *B3GLCT* gene (formerly known as *B3GALTL*) provides instructions for making an enzyme called beta 3-glucosyltransferase (B3Glc-T), which is involved in the complex process of adding sugar molecules to proteins (glycosylation). Glycosylation modifies proteins so they can perform a wider variety of functions. The B3Glc-T enzyme is involved in a two-step glycosylation pathway that results in the formation of a sugar structure, made up of the sugars fucose and glucose, on a specific location of several different proteins. The B3Glc-T enzyme is responsible for the second step, which adds a glucose molecule to the fucose molecule already attached to the protein. The *B3GLCT* gene is normally turned on (active) in most cells of the body, which suggests that the B3Glc-T enzyme plays an important role across many cell types.

Health Conditions Related to Genetic Changes

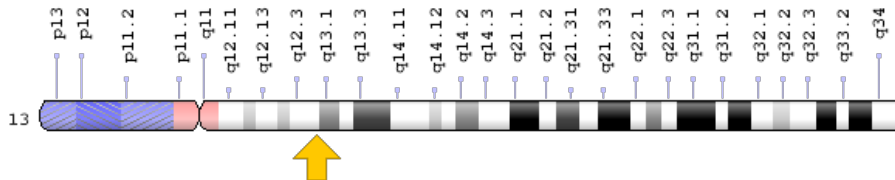
Peters plus syndrome

At least 10 mutations that cause Peters plus syndrome have been identified in the *B3GLCT* gene. Peters plus syndrome is characterized by eye abnormalities, short stature, intellectual disability, and distinctive facial features. The most common *B3GLCT* gene mutation replaces the DNA building block (nucleotide) guanine with the nucleotide adenine near an area of the gene called exon 8 (written as 660+1G>A). This mutation disrupts how genetic information is pieced together to produce the B3Glc-T enzyme. The resulting enzyme is abnormally short and nonfunctional. It is unclear how the loss of functional B3Glc-T enzyme leads to the signs and symptoms of Peters plus syndrome, but impaired glycosylation likely disrupts the function of many proteins, which may contribute to the variety of features.

Chromosomal Location

Cytogenetic Location: 13q12.3, which is the long (q) arm of chromosome 13 at position 12.3

Molecular Location: base pairs 31,199,975 to 31,332,276 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- B3GALT
- B3Glc-T
- B3GLT_HUMAN
- B3GTL
- beta 1,3-galactosyltransferase-like
- beta-3-glycosyltransferase-like
- beta3Glc-T

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Glycosyltransferases and Glycan-processing Enzymes
<https://www.ncbi.nlm.nih.gov/books/NBK1921/>
- Molecular Biology of the Cell (fourth edition, 2002): What is the Purpose of Glycosylation?
<https://www.ncbi.nlm.nih.gov/books/NBK26941/#A2354>

GeneReviews

- Peters Plus Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1464>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28B3GALTL%5BTIAB%5D%29+OR+%28beta1,3-galactosyltransferase%5BTIAB%5D%29+OR+%28B3GLCT%5BTIAB%5D%29+OR+%28beta+3-galactosyltransferase%5BTIAB%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- BETA-3-GLUCOSYLTRANSFERASE
<http://omim.org/entry/610308>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=B3GLCT%5Bgene%5D>
- HGNC Gene Family: Beta 3-glycosyltransferases
<http://www.genenames.org/cgi-bin/genefamilies/set/426>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20207
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/145173>
- UniProt
<http://www.uniprot.org/uniprot/Q6Y288>

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